A polymorphic DNA sequence from the terminal part of chromosome 20q [D20S26]

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Source/Description: The probe IP20K09 is a 12 kb SalI fragment isolated from a λ phage obtained by screening a human genomic library with a STIR repeated element (1). The insert is subcloned in the SalI site of pBS+ vector (Stratagene).

Polymorphism: TaqI identifies 3 alleles defined by a 5.8 kb fragment, a 5.4 kb fragment and a 3.3 kb + 2.1 kb doublet. Several other non polymorphic fragments are also detected.

Frequency: Estimated from 46 parents of the CEPH panel.

5.8 kb allele: 0.10 5.4 kb allele: 0.47 3.3 + 2.1 kb allele: 0.43

Not Polymorphic For: BamHI, BglII, EcoRI, HindIII, MspI, PstI, RsaI, XbaI (studied in 5 unrelated individuals).

Chromosomal Localization: Localized to 20q13.3 by in situ hybridization and genetic linkage analysis (1).

Mendelian Inheritance: Codominant segregation observed in 20 CEPH families.

Probe Availability: The probe will be submitted to the American Tissue Culture Collection (ATCC). Until it is made available through the ATCC it can be obtained from J. Weissenbach.

Other Comments: Segregation data included in the CEPH database version 3. RFLPs are observed under normal hybridization and wash (0.2×SSC) stringencies.

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Reference: (1) Rouyer, F. et al. (1990) EMBO J. 9, 2: 505-514.

A new polymorphic probe on chromosome 3p: λLIB45 - 82 (D3S232)

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Source/Description: λLIB45-82 is a 2.2 kb EcoRI fragment isolated from a Charon 21A human chromosome 3 library (1).

Polymorphism: MspI digestion of genomic DNA and hybridization with the probe detects a two allele polymorphism: 4.5 kb (M1) and 3.45/1.05 kb (M2). No constant bands were present.

Frequency: Estimated from 36 unrelated Caucasians.

M1:0.64M2:0.36

Not Polymorphic For: BamHI, BglII, DraI, EcoRI, HindIII, Hinfl, PvuII, RsaI and TaqI.

Chromosomal Localization: Using a somatic cell hybrid panel (2) which was based on linkage groups anchored to physically localized markers, the probe was assigned to the short arm of chromosome 3 between 3p21 and 3pter.

Mendelian Inheritance: Mendelian inheritance has been demonstrated in two three-generation von Heppel-Lindau disease (vHL) families.

Probe Availability: Available for collaboration.

Other Comments: The probe sequences are highly conserved between human and rodent species.

References: 1. American Type Culture Collection No. 57748, National Lab ID code LA03NS02. 2. Brauch, H. et al. (1989) A somatic cell hybrid panel for physical assignment of 3p probes. Abstract, HGM10. Cytogenet. Cell Genet. 51(1-4), 968.

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